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Congenital Tritanopia: A Comparison of Two Clinical Cases

Description

This poster discusses two patient cases of presumed blue-yellow color deficiency in order to help optometrists care for this rare class of patients. Since this condition is extremely uncommon at birth, and is not possible to see by looking in the eye, practitioners may miss the symptoms of this condition. Optical treatment options are also discussed.

Disciplines

Optometry

Comments

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CONGENITAL TRITANOPIA: A COMPARISON OF TWO CLINICAL CASES

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ABSTRACT

This poster explores two cases of presumed blue-yellow color deficiency in order to help optometrists care for this rare class of patients. Since this condition is extremely uncommon at birth, and is not possible to see by looking in the eye, practitioners may miss the subtle signs and less subtle symptoms. Optical treatment options are also discussed.

SUBJECTIVE FINDINGS

Congenital tritan color vision defects are an autosomal dominant genetic condition affecting 0.005% of Caucasian males. This translates to one in 20,000 patients. Two suspected blue-yellow color deficient cases are discussed below.

Patient #1 was a 32-year-old white male with a chief complaint of increased sensitivity to lights in both eyes, and headaches. Wearing sunglasses over his contact lenses was shown to reduce symptoms, but on further questioning, the patient reported substantial photophobia even with sunglasses. Ocular and medical histories were unremarkable, except the patient reported smoking less than one pack of cigarettes per day.

Patient #2 was a 27-year-old white male. His concern was difficulty seeing in bright light, and especially when facing the setting sun. The problem was in both eyes, stable more or less since birth, relieved slightly by anti-reflective coating and Transitions photochromic lenses. His difficulties were severe. Under the conditions described above he can only see six inches in front of his face, though with Transitions in his glasses his range is extended to three feet.

Patient #2 reports that he is missing sensitivity to blue, which looks gray to him. He has a very difficult time reading e-books, though does better on Kindles. His computer is always on the dimmest illumination, his house is constantly blacked out with shades, and he uses 60W bulbs or less. He states that he has above-average night vision. He relies mainly on his hearing to cross intersections without getting hit.

Patient #2 had a normal birth as far as he knows, without supplemental oxygen or forceps. He reports his nutrition to be good, and his systemic health unremarkable, not taking any medications.

Patient #2 reported that his father has the same set of symptoms, and likely his PGF as well. He has been seen by three eye care physicians already and has been told that he was “making things up.”

Patient #2 reported that he was a direct descendant of the House of Stewart, and he states that his lack of blue sensitivity goes back generations and includes Bonnie Prince Charlie (Figure 1).



Figure 1: Charles Edward Stuart, or “Bonnie Prince Charlie,” who may have passed on the tritan gene to patient #2

OBJECTIVE FINDINGS

For patient #1, refractive testing revealed low myopia and 6^A esophoria at near. A full exam and color testing was performed using the Optec 2000 and the patient scored a 3/5, and a 2/5 on repeat testing. Note that this color vision test is for red-green defects.

Patient #2 had a manifest Rx: OD -5.00 -0.50 x 160 VA 20/15, OS -5.75 DS VA 20/20+1. Extraocular motilities were full, as was a screening visual field. Pupils were large, but equal, round and reactive to light, with no afferent pupillary defect. Intraocular Pressures were 15/15 @ 9:50 AM with an iCare tonometer.

Patient #2 was also tested with Short-Wavelength Automated Perimetry, also known as SWAP, or blue-yellow visual fields, which showed moderate depression in the mid-peripheral. Scanning retinal laser in the form of OCT (Optical Coherence Tomography) showed no thinning of any of the retinal layers. A retinal fundus photo and Cirrus OCT of the right eye is shown in Figure 2 and Figure 3.

ASSESSMENT & PLAN



Figure 2: Right retina of patient #2. OS was similar.

Patient #1 was diagnosed photophobia secondary to moderate to severe tritanomaly. This condition was presumed to be congenital, and was likely contributing to his headaches. Congenital color vision deficiencies are not expected to progress.

For patient #1, because standard sunglasses did little to alleviate symptoms and caused too much disruption with indoor activities, varying levels of blue tint was trial framed, and blue #1 was the preferred tint by the patient. The patient came for a CVE two years later and was enjoying comfortable vision using the blue #1 tint on his lenses. At this appointment, the patient reported being diagnosed with transverse myelitis of the spine. This kind of inflammation can lead to demyelination, but is not expected to affect vision.

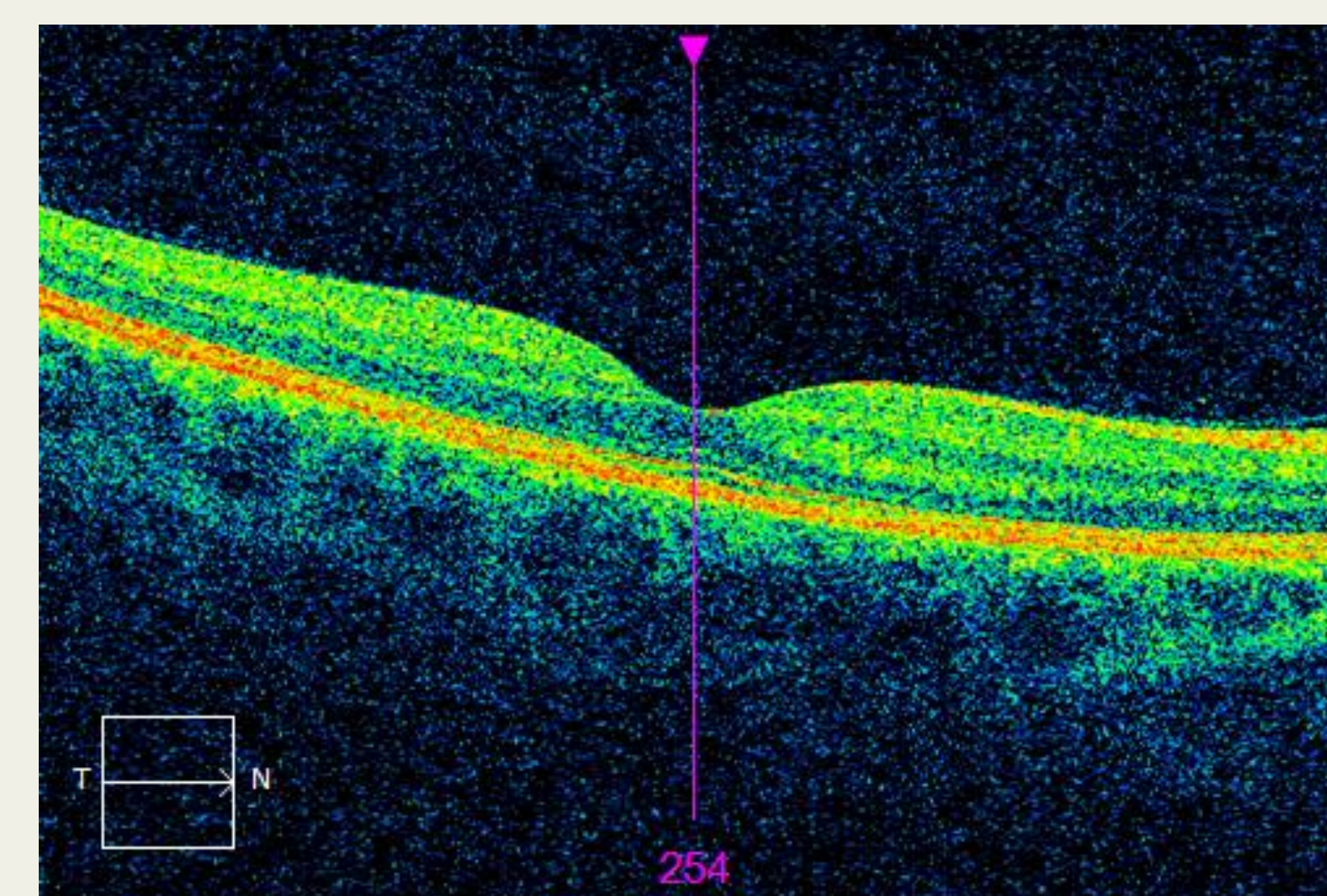


Figure 3: Right Cirrus OCT of patient #2. OS was similar.

Patient #2 was diagnosed with mild to moderate congenital tritanomaly. He was educated about the genetic nature of his condition, which was supported by his family history. He did not request any treatment beyond his habitual prescription, but based on the treatment for patient #1, blue #1 tint was recommended, as detailed above.

CONCLUSIONS

For tritan patients, we suspect that the blue lenses acted as a neutral density (or gray) filter for the patient. This is because tritans see gray at 380 nm, which is the violet end of the visible spectrum.

For diagnosis, we recommend the following testing:

- Hardy-Rand-Rittler (HRR) testing is recommended, which includes screening and diagnostic pseudoisochromatic plates for mild, moderate, and severe tritan defects.
- When available, SWAP (Short-Wavelength Automated Perimetry), or blue-yellow visual fields may yield additional information (see Figure 4, below).

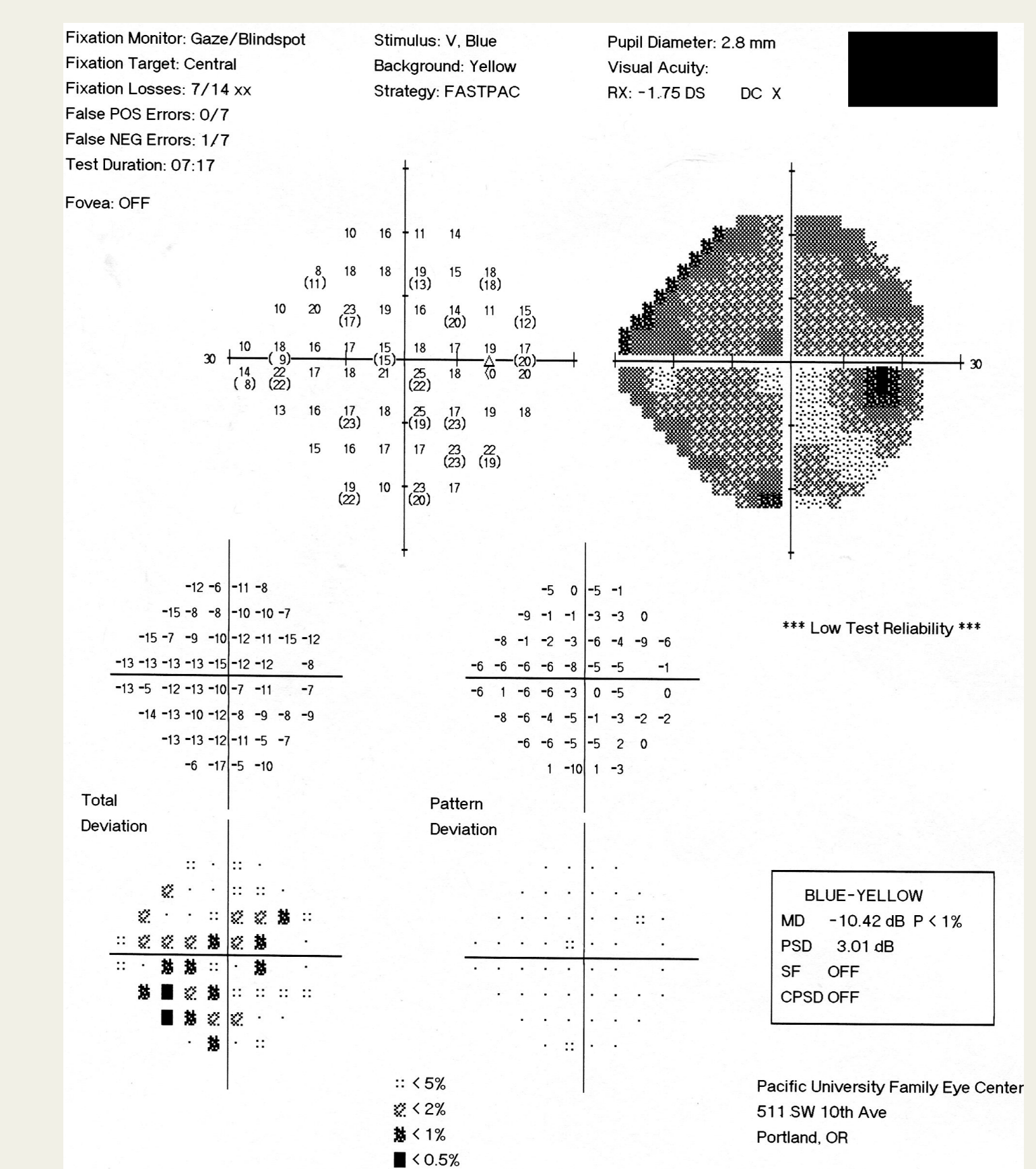


Figure 4: SWAP (Blue-Yellow) Visual Field for the right eye of Patient #2. OS was similar.

For treatment, we recommend a blue tint to improve visual comfort. Blue-blocker (yellow) tints would we predicted to make their vision less comfortable, as was proven to be the case with patient #1.

LITERATURE CITED

1. http://en.wikipedia.org/wiki/Charles_Edward_Stuart
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